SLC2A10 Gene

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solute carrier family 2 member 10

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1. Normal Function

The *SLC2A10* gene provides instructions for making a protein called GLUT10. GLUT10 is classified as a glucose transporter; this type of protein moves the simple sugar glucose across cell membranes and helps maintain proper levels of glucose within cells. However, GLUT10 has some structural differences from other glucose transporters, and its role in the movement of glucose or other substances is unclear.

The level of GLUT10 appears to be involved in the regulation of a process called the transforming growth factor-beta (TGF-β) signaling pathway. This pathway is involved in cell growth and division (proliferation) and the process by which cells mature to carry out special functions (differentiation). The TGF-β signaling pathway is also involved in bone and blood vessel development and the formation of the extracellular matrix, an intricate lattice of proteins and other molecules that forms in the spaces between cells and defines the structure and properties of connective tissues. Connective tissue provides strength and flexibility to structures throughout the body, including blood vessels, skin, joints, and the gastrointestinal tract.

Studies indicate that GLUT10 may also be involved in the functioning of mitochondria, the energy-producing centers within cells.

2. Health Conditions Related to Genetic Changes

2.1. Arterial tortuosity syndrome

At least 23 *SLC2A10* gene mutations have been identified in people with arterial tortuosity syndrome, a connective tissue disorder characterized by abnormal curving and twisting (tortuosity) of the blood vessels that carry blood from the heart to the rest of the body (arteries) and other health problems.

The mutations that cause arterial tortuosity syndrome reduce or eliminate GLUT10 function. By mechanisms that are not well understood, a lack (deficiency) of functional GLUT10 protein leads to overactivity (upregulation) of TGF-β signaling. Excessive growth signaling results in elongation of the arteries. Since the end points of the arteries are fixed, the extra length twists and curves, leading to tortuosity. Overactive TGF-β signaling also interferes with normal formation of the connective tissues in other parts of the body, leading to the joint and skin abnormalities and other features of arterial tortuosity syndrome.

Changes in mitochondrial function related to GLUT10 deficiency may also affect cardiovascular system development, but the relationship between mitochondrial function and the specific signs and symptoms of arterial tortuosity syndrome is unclear.

2.2. Other disorders

Several normal variations (polymorphisms) of the *SLC2A10* gene have been associated with an increased risk of peripheral artery disease in people with type 2 diabetes, a disorder in which resistance to the hormone insulin leads to excess glucose levels in the blood (hyperglycemia). Peripheral artery disease is a condition in which an accumulation of fatty deposits and scar-like tissue in the lining of the arteries (atherosclerosis) reduces blood flow to the legs, causing pain

when walking. Problems with blood vessels, including peripheral artery disease, are common in type 2 diabetes, and are believed to be related to the effect of hyperglycemia on TGF-β signaling. Alterations in the GLUT10 protein caused by *SLC2A10* gene variations may also affect TGF-β signaling and increase the risk of blood vessel problems in diabetes.

3. Other Names for This Gene

- ATS
- glucose transporter type 10
- GLUT-10
- GLUT10
- GTR10_HUMAN
- solute carrier family 2 (facilitated glucose transporter), member 10
- solute carrier family 2, facilitated glucose transporter member 10

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